Crouzon syndrome

Crouzon syndrome is a genetic disorder characterized by the premature fusion of certain skull bones (craniosynostosis). This early fusion prevents the skull from growing normally and affects the shape of the head and face.

Many features of Crouzon syndrome result from the premature fusion of the skull bones. Abnormal growth of these bones leads to wide-set, bulging eyes and vision problems caused by shallow eye sockets; eyes that do not point in the same direction (strabismus); a beaked nose; and an underdeveloped upper jaw. In addition, people with Crouzon syndrome may have dental problems and hearing loss, which is sometimes accompanied by narrow ear canals. A few people with Crouzon syndrome have an opening in the lip and the roof of the mouth (cleft lip and palate). The severity of these signs and symptoms varies among affected people. People with Crouzon syndrome are usually of normal intelligence.

Frequency

Crouzon syndrome is seen in about 16 per million newborns. It is the most common craniosynostosis syndrome.

Genetic Changes

Mutations in the *FGFR2* gene cause Crouzon syndrome. This gene provides instructions for making a protein called fibroblast growth factor receptor 2. Among its multiple functions, this protein signals immature cells to become bone cells during embryonic development. Mutations in the *FGFR2* gene probably overstimulate signaling by the FGFR2 protein, which causes the bones of the skull to fuse prematurely.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Craniofacial dysarthrosis
- Craniofacial Dysostosis
- Craniofacial dysostosis syndrome
- Craniofacial dysostosis, type 1; CFD1
- Crouzon craniofacial dysostosis

- Crouzon's Disease
- Crouzons Disease

Diagnosis & Management

These resources address the diagnosis or management of Crouzon syndrome:

- GeneReview: FGFR-Related Craniosynostosis Syndromes
  https://www.ncbi.nlm.nih.gov/books/NBK1455
- Genetic Testing Registry: Crouzon syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0010273/
- MedlinePlus Encyclopedia: Craniosynostosis
  https://medlineplus.gov/ency/article/001590.htm

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Craniosynostosis
  https://medlineplus.gov/ency/article/001590.htm
- Health Topic: Craniofacial Abnormalities
  https://medlineplus.gov/craniofacialabnormalities.html

Genetic and Rare Diseases Information Center

- Crouzon syndrome
  https://rarediseases.info.nih.gov/diseases/6206/crouzon-syndrome

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Craniosynostosis Information Page
  https://www.ninds.nih.gov/Disorders/All-Disorders/Craniosynostosis-Information-Page

Educational Resources

- Boston Children's Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/crouzon-syndrome
- Collaboration for Craniofacial Development and Disorders, Johns Hopkins Medicine
  http://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/pediatric_neurosurgery/conditions/craniosynostosis/
- Disease InfoSearch: Crouzon Syndrome
  http://www.diseaseinfosearch.org/Crouzon+Syndrome/2018
- MalaCards: crouzon syndrome
  http://www.malacards.org/card/crouzon_syndrome
- Orphanet: Crouzon disease
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=207
- Seattle Children's Hospital and Regional Medical Center
  http://www.seattlechildrens.org/medical-conditions/chromosomal-genetic-conditions/crouzon-syndrome/
- UC Davis Children's Hospital
  http://www.ucdmc.ucdavis.edu/children/clinical_services/cleft_craniofacial/anomalies/crouzon.html

Patient Support and Advocacy Resources

- Children's Craniofacial Association
  http://www.ccakids.com
- Cleft Palate Foundation
  http://www.cleftline.org/parents-individuals/publications/crouzon-syndrome/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/crouzon-syndrome/
- Resource List from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/craniofa.html

GeneReviews

- FGFR-Related Craniosynostosis Syndromes
  https://www.ncbi.nlm.nih.gov/books/NBK1455

Genetic Testing Registry

- Crouzon syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0010273/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22crouzon+syndrome%22+OR+%22craniofacial+abnormalities%22+OR+%22craniofacial+dysostosis%22

Scientific articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Craniofacial+Dysostosis%5BMAJR%5D%29+AND+%28Crouzon+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- CROUZON SYNDROME
  http://omim.org/entry/123500

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC38845/
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